



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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O.G. FIG.			
APPROVED	BY	CLASS	SUBCLASS
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Chromosome 13

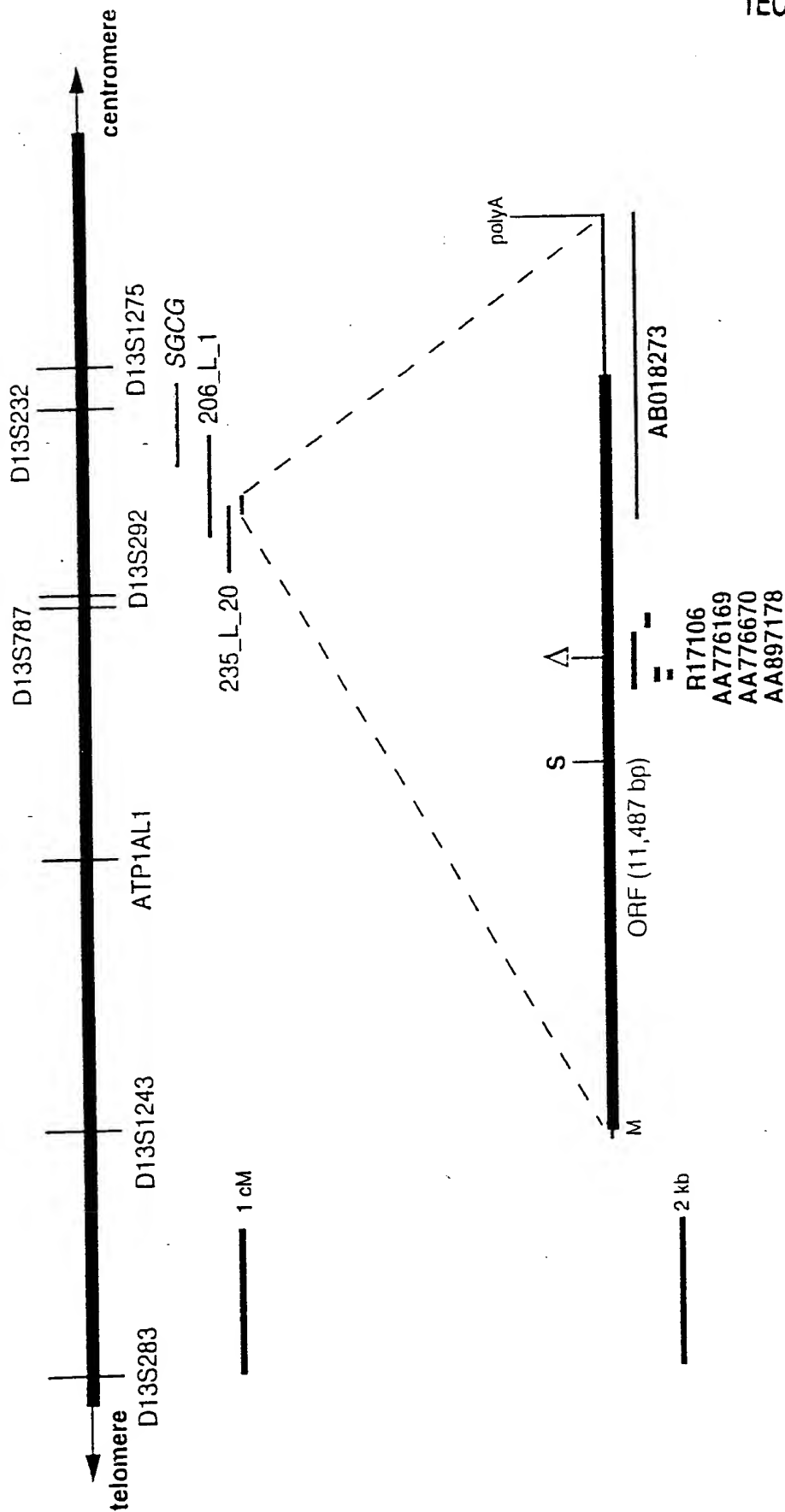


Figure 1



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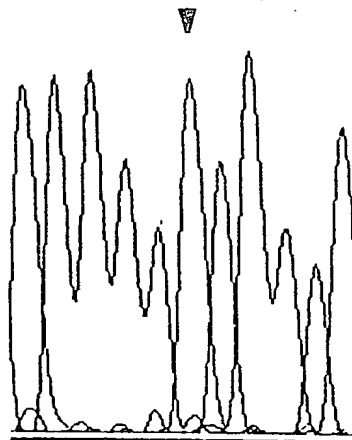
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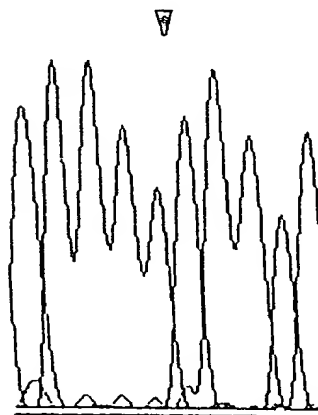
APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

ACCCTATTCA
(SEQ ID NO: 17)



6594T

ACCCTATTCA
(SEQ ID NO: 18)



6594Δ

Figure 2A



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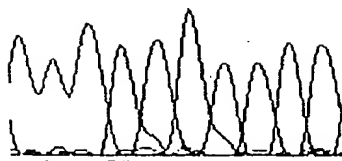
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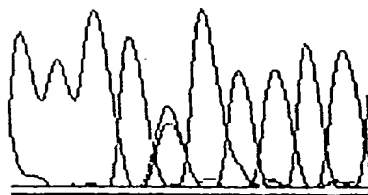
APPROVED BY	CLASS	SUBCLASS
	DRAFTSMAN	

A A A G C G A C A C
(SEQ ID NO: 19)



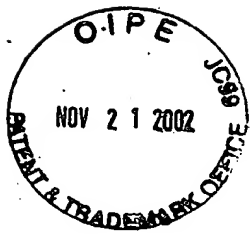
5254C

A A A G N G A C A C
(SEQ ID NO: 20)



5254C/T

Figure 2B



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	CLASS	SUBCLASS

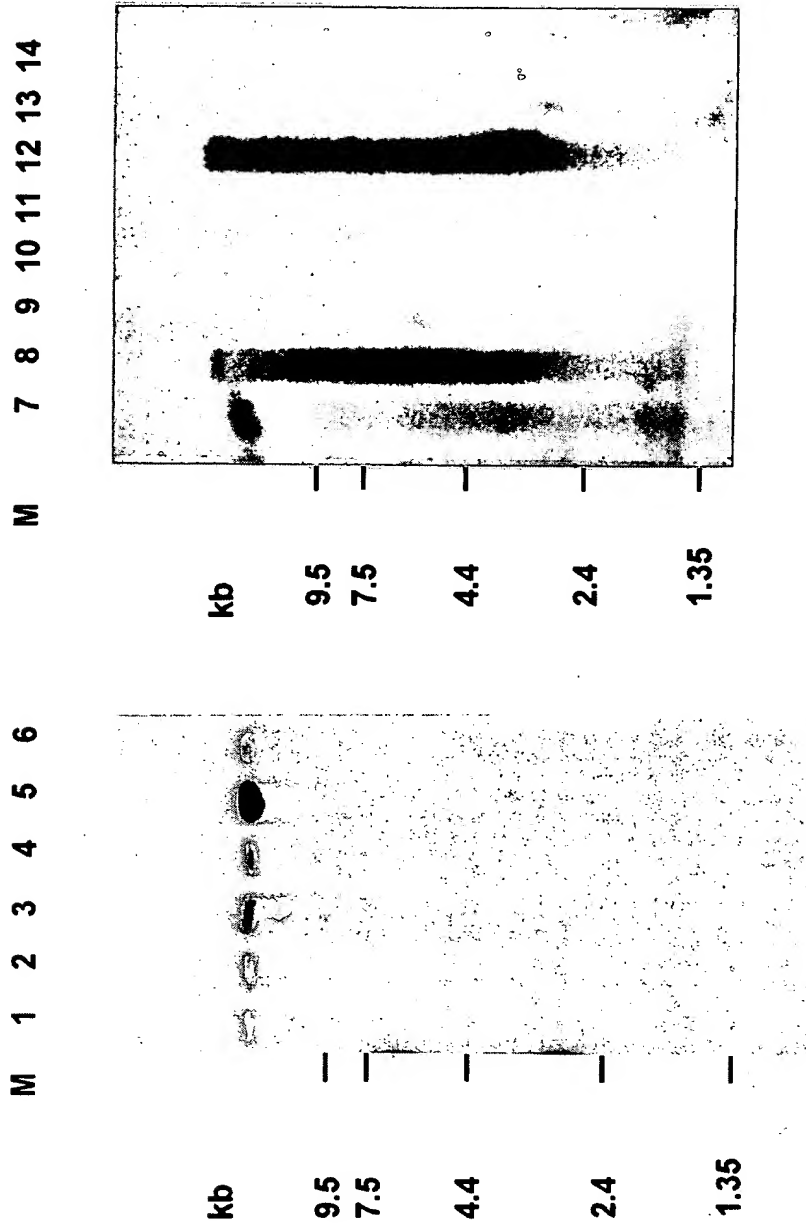


Figure 3

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APPROVED		O.G. FIG.	
BY	CLASS	BY	SUBCLASS
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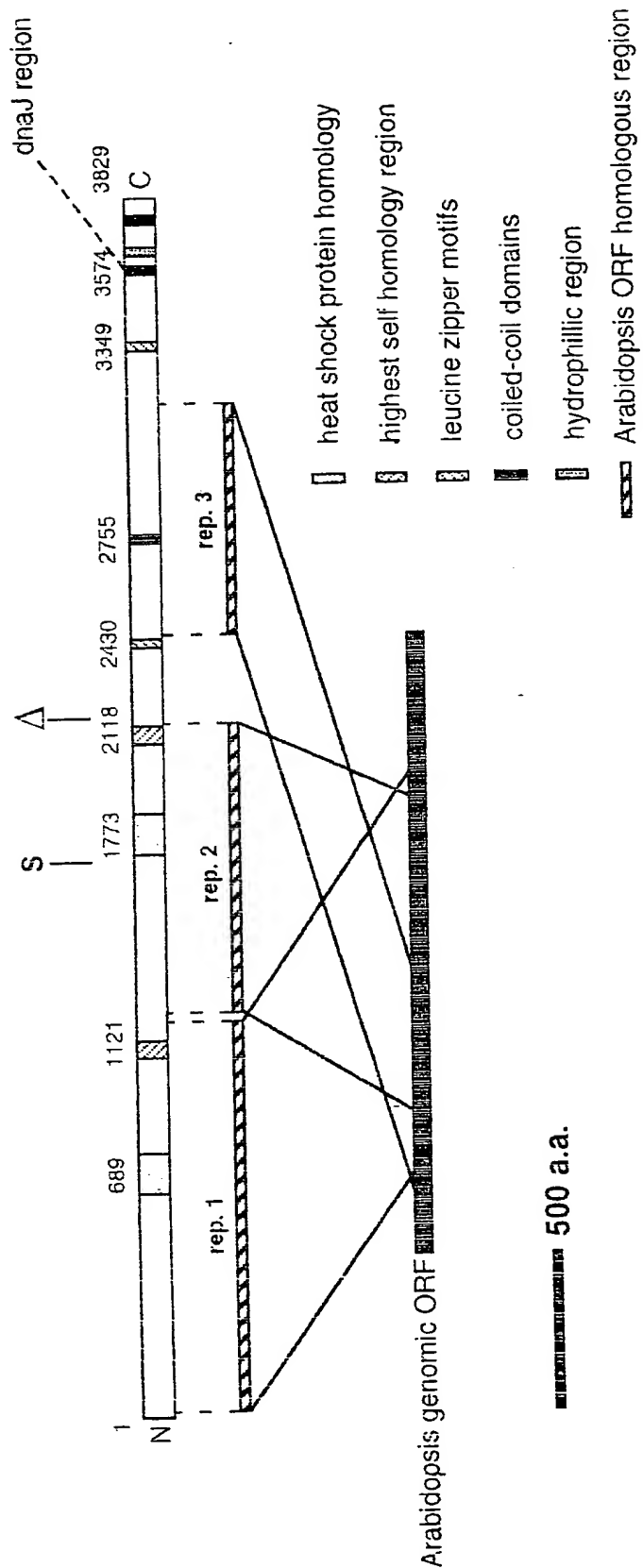


Figure 4A



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APPROVED	O.G. FIG.	
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HUMAN-REP 1	705	GOREPLTVRIKNILEEYPSVSDIFKEILQONADOAN-ATECSFLIDMRNMDIRENLLDPG
MOUSE-REP 1	705	GOREPLTVRIKNILEEYPSVSDIFKEILQONADOAN-ATECSFLIDMRNMDIRENLLDPG
HUMAN-REP 2	1773	Gqke.LT.RIK.IL.YPS.em.keILQONADOA..ATE..Fv.D.R.....d.i.D..
ARABIDOPSIS	987	Gqhe.LT.RIK.ILE.Y.....I..EIYQNAEOA..ASE.tFLID.h.....d.LL.P.
HSP90 (YEAST)	18	.Q.E..l....I...Y.....rELi.NA.OA.....L.D.k.....d.i....
HUMAN-REP 1	764	MAACHGPALWSFNNSQ-FSDSDFIN----ITRLG-----ESLKRGE-VDKVGKEGLGFNS
HUMAN-REP 1	764	MAACHGPALWSFNNSQ-FSDSDFIN....ITRLG.....ESLKRGE-VDKVGKEGLGFNS
HUMAN-REP 2	1830	.A...GPAL..yNN...Fte.D.....I..LG.....K.G....K.G.YGIGFNS
ARABIDOPSIS	1044	MA...GPALy.FNNS..Fe.D.....IsriG.....qa.K.....igrPGLGFN.
HSP90 (YEAST)	78L...q.....L.ae.in....I.k.G.....EaL..G..V..iG.FGYGF.S
HUMAN-REP 1	813	VYHITDIPiIMSR-----EFMIMFDP
MOUSE-REP 1	813	VYHITDIPiIMSR.....EFMIMFDP
MOUSE-REP 2	1879	VYHITD.P..iS.....d.l.iFD.
ARABIDOPSIS	1093	VYH.TDIP..vS.....E.ivMFDP
HSP90 (YEAST)	137	If.v.D...visk.....F.v..D.

Underline = conserved
Bold = identical

Figure 4B



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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

MNTFWPGRELIVQWYPFDENRNHPSVSWLKMVWKNLYIHFSEDLTFLFDEMPLIPRTILEEV.....S.DKR...L.....L.N.	60
GQTCVELIRLRIPSLVILDDESEALPEFLADIVQKLGGFVLKKLDASIQHPLIKKYIHS D.....V.....T.....I...R...T.....V.....	120
PLPSAVLQIMEKMPLQKLCNQITSLLPTHKDALRKFLASLTDSSSEKEKRIIQELAIFKRII.....I.....A.....T.....T.....	180
NHSSDQGISSYTKLKGCKVLHHTAKLPADLRLSISVIDSSDEATIRLANMLKIEQLKTTSD.....T.....V.....K.....	240
CLKLVLKDIENAFYSHEEVTLQMLWVLENLSSLKNENPNVLEWLTPLKFIQISQEQMVSA ...F....G....TQ.....I.....S...D..M....HM..GHV.A.	300
GELFDPDIEVLKDLFCNEEGTYFPSPVFTSPDILHSLRQIGLKNEASLKEKDVVQVAKKI .D.....R...Y...EAC...TI.....S.....R..	360
EALQVGACPDQDVLKKAKTLLLVLNKNHTLLQSSEGKMTLKKIKWVPACKERPPNYPGSSS.QN....M.....Q.....A.....	420
LVWKGDLNLCAPPDMCDVGHAILIGSSSLPLVESIHVNLEKALGIFTKPSLSAVLKHFKIAA..V.V.....V....Q..S....TIN.....T	480
VVDWYSSKTFSDEDYYQFHILLEIYGFMHDLNEGKDSFRALKFPWVWTGKKFCPLAQT.....S.....K.....N.....	540
VIKPIHDLDLQPYLHNVPKTMKFHQLFKVCGSIEELTSDHISMVIQKIYKSDQDLSEQ ...T.....Y.....A.....V.....E....E	600
ESKQNLHMLNIIRWLYSNQIPASPNTVPPIHHSKNPSKLIMKPIHECCYCDIKVDDLNDM.....Y..R....V.....	660
LLEDSEVEPIILVHEDI PMKTAEWLKVPCLSLRLINPENMGFEQSGQREPLTVRIKNILEE	720
YPSVSDIFKELLQNADDANATECSFLIDMRRNMDIRENLLDPGMAACHGPALWSFNNSQFM.....E.	780
SDSDFVNITRLGESLKRGEVDKVGKFGGLGFNSVYHITDIPIMMSREFMIMFDPNINHISKL.....	840
HIKDKSNPGIKINWSKQKRLRKFPNQFKPFIDVFGCQLPLTVEAPYSYNGTLFRLSFRT ...R.....A.....	900
QQEAKVSEVSSTCYNTADIYSLVDEFSLCGHRLIIFTQSVKSMYLYKIEETNP SLAQDN.....	960
TVI IKKSCSSKALNTPVLSVLKEAAKLMKTCSSSNKKLPSDEPKSSCILQITVEEFHHV .I....V.P....A.....T.V.....	1020
FRRIADLQSPLFRGPDDPAALFEMAKSGQSKKPSDELSQKTVECTTWLLCTCMDTGEALT.....P....D....I.....	1080
KFSLSESGRRLGLVPCGAVGVQLSEIQDQKWTVKPHIGEVFCYLPRIKTGLPVHINGCF ...N.....L.H.T.E.....I.....	1140
AVTSNRKEIWKTDTKGRWNTTFMRHVIVKAYLQVLSVLRDLATS GELMDYTTYAVWPD PDA.....IG...T.....	1200
LVHDDFSVICQGFYEDIAHGKGKELTKVFS DGSTWVSMKNVRLDDSILKRRDVGSAAFKK.....R.....M.....Q.K.....	1260
IFLKYLKKTGSKNLCAVELPSSVKLGFE EAGCKQILLENTFSEKQFFSEVFFPNIQEIEAA.....	1320
ELRDPLMIFVLNEKVDEFSGVLRVTPCIPCSLEGHPLVLP SRLIHPEGRVAKLFDIKDGRN.....L....I.....V.....T.....	1380
FPYGSTQDYLNPIILI KLVLGKAKDDILWDDMLERAVSVAEINKSDHVAACL RSSILLSM.....E.....A.....	1440

Figure 5A



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APPROVED	O.G. FIG.	
	BY	DRAFTSMAN
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LIDEKLRKIRDPRAKDFAAKYQTIRFLPFLTKPAGFSLDWKGNFSFKPETMFAATDLYTAEHK.....P.....E.....I.....Y	1500
QDIVCLLQPILNENSHSFRGCGSVSLAVKEFLGLLKPTVDLVINQLKEVAKSVDDGITLQ.....	1560
YQENITNACYKYLHEALMQNEITKMSIIDKLPFSFILVENAYVDSEKVSFHLNFEAAPYVL...MA.AT..E.....C.....V..E.....	1620
LYQLPNKYKNNFRELFTVGVRSCTVEDFALVLESIDQERGTKQITEENFQLCRRIISES.....F.....K.....	1680
GIWSLIREKKQEFCEKNYKILLPDTNLMLLPAKSLCYNDPCWIKVKDTTVKYCHADIPRR.....L.....S.....	1740
EVAVKLGAVPKRHKALERYASNVCFTTLGTEFGQKEKLTSRIKSILNAYPSEKEMLKELLI.....I...A.....	1800
QNADDAKATEICFVFDPRQHPVDRIFFDDKWAPLQGPALCVYNNQPFTEDDVRGIQNLGKG	1860
TKEGNPYKTGQYGIGFNSVYHITDCPSFISGNDILCIFDPHARYAPGATSISPGRMFRDLC...H.....G.....V.....	1920
DADFRTQFSDVLDLYLGTHFKLDNCTMFRFPLRNAEMAKVSEISSVPASDRMVQNLLDKLN.....Q.....S.....	1980
RSDGAELLMFLNHMEKISICEIDKSTGALNVLYSVKGKITDGDRLKRKQFHASVIDSVTKA..G.....	2040
KRQLKDIPVQQITYTMDTEDSEGNLTTWLICNRSGFSSMEKVSKSVISAHKNQDITLFPR	2100
GGVAACITHNYKKPHRAFCFLPLSLETGLPFHVNGHFALDSARRNLWRDDNGVGVRSDWN	2160
NSLMTALIAPAYVELLIQLKKRYFPGSDPTLSVLQNTPIHVVKDTLKKFLSFFPVNRLDL	2220
QPDLYCLVKALYNCIHEDMKRLLPVVRAPNIDGSDLHSAVITWINMSTSNKTRPFFDNLS.....	2280
LQDELQHLKNADYNITTRKTVAENVYRLKHLLEIGFNLVYNCDETANLYHCLIDADIPVV.....	2340
SYVTPADIRSFLMTFSSPDNTCHIGKLPCLQQTNLKLHSLKLLVDYCFKDAEENEIEVV.....S.F..	2400
EGLPLLITLDSVLQTFDAKRPKFLTTYHELIPSRKDLFMNTLYLKYSNILLNCKVAKVFDI..G.....SV.....	2460
ISSFADLLSSVLPREYKTKSCTKWKDNFASESWLKNAWHFISESVSVKEDQEETKPTFDIN.A.....TD...P..A..V	2520
VVDTLKDWALLPGTKFTVSANQLVVPEGDVLLPLSLMHIAVFPNAQSDKVFHALMKAGCI I..I.....TS.....I.....	2580
QLALNKICSKDSAFVPLLSCHTANIESPTSILKALHYMVQTSTFRAEKLVENDFEALLMYL.....D..A.....V.....T...M.....	2640
FNCNLNHLMSQDDIKILKSLPCYKSISGRYVSIGKFGTCYVLTKSIPSAEVEKWTQSSSSS.....M..A.....	2700
AFLEEKIHLKELYEVIGCVPVDDLEVYLKHLPLKIENTLSYDAKLEHLIYLNRLSSAEELV.....L.....A.I..P	2760
<u>SEIKEQLFEKLESLLIIHDANSRLKQAKHFYDRTVRVFEVMLPEKLFIPNDFFKLEQLI</u>N.....KE.....V.	2820
KPKNHVTFMTSWVEFLRNIGLKYILSQQLLQFAKEISVRANTENWSKETLQNTVDILLHQAA.....A.....S.....	2880

Figure 5B



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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
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HI FQERMDLLSGNFLKELSLIPFLC PERAPAEFIRFHPQYQEVNGTLPLIKFNGAQVNP	2940
.....Y.....	
FKQCDVLQLLWTSCPI LPEKATPLS I KEQEGSDLG PQEQLEQVLNMLNVNLD PPLDKVIN	3000
.....A.....	
NCRNICNITTLDEEMVKTRAKVLR SIYEFLSAEKREFRFQLRGVAFVMVEDGWKLLKPEE	3060
.....	
VVINLEYESDFKPYLYKLPLELGT FHFQLFKHLGTEDIISTKQYVEVLSRIFKNSE GKQLD	3120
.....A.....S.....	
PNEMRTVKRVVSGLFRSLQND SVKVRSDLENVRDLALYLP SQDGRLVKSSILVFDDAPHY	3180
.....K.....A.....K.....	
KSRIQGNIGVQMLVDLSQC YLGKDHGFHTKLIMLFPQKLRPRLSSILEEQ LDEETPKVC	3240
.....	
QFGALCSLQGR LQLLSSEQFITGLIRIMKHENDNAFLANE EKAIRLCKALREGLKVSCF	3300
.....	
EKLQTTLRVKGFNPIPHSRSETFA FLKRFGNVILLYIQHSDSKDINFL LALAMTLKSAT	3360
.....	
DNLISDTSYLIAMLCNDIYRIG EKLDLGVKYDSSEPSKLELPMPTPIPAEIH YTLML	3420
.....S.....	
DPMNVFYPGEYVGYLVDAE GGDYGSYQPTYTYAIIVQEVEREDADN SSFLGKIYQIDIG	3480
.....T.....	
YSEYKIVSSLDLYKFSRPE ESSQSRDSAPSTPTSPTEFLT PGLRSIPPLFSGRESHKT-S	3540
.....D...N...T.....K...SP.	
SKHQSPKKLVNSLPEILKEVTS VVEQAWKL PESERKKI IRRLYLKWHPDKNPFN HDIAN	3600
T..H..R....A.....	
<u>EVFKHLQNEINRLEKQAF LQADNRASRTFSTSASRFQSDKYSFQRFYTSWNQEATSHK</u>	3660
.....	
<u>SERQQONKEKCPPSAGQTY SQRFVPPTFKSVGNPVEARRWLRQARANFSAARNDLHKNA</u>	3720
.....S.....	
NEWVCFKCYLSTKLALIAADYAVRGKSDKDV KPTALAQKIEEYSQOLEGLTNDVHTLEAY	3780
.....	
GVDSLKTRYPDLLPFPQIPNDRFTSEVAMRVMECTACII IKLENFMQQKV	3830
.....I.....	

Figure 5C

APPROVED	O.G. FIG.	
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Table 1 ESTs identified by sample-sequencing of the ARSACS critical interval

BAC clone	GenBank #	UniGene	Identity*	Tissue Source
235_I_20	AA987300	IIs. 129092	221/230	neuroendocrine lung carcinoids
235_I_20	AA476635*		249/296	total fetus
235_I_20	AI351876*		272/335	melanocyte, fetal heart, pregnant uterus (pool)
235_I_20	W25994	IIs. 163732	447/464	retina
235_I_20	AI377467	IIs. 163732	257/263	total fetus
235_I_20	AA601007		488/491	schwannoma tumor
235_I_20	AA324964		214/234	cerebellum
235_I_20, 206_I_1	AA897178 ^b		238/238	fetal lung, testis, and B-cell
235_I_20, 206_I_1	RI7106 ^b	IIs. 188560	747/784	brain, adipose tissue
235_I_20, 206_I_1	AB018273 ^b	IIs. 159492	4318/4318	multiple tissue types including brain, CNS, and whole embryo

a: Number of homologous nucleotides between the BAC subclone sequence and the GenBank sequence.

b: AA897178, RI7106, and AB018273 are all contained within *spatin*.

*: AA176335 and AI351876 were homologous to non-overlapping portions of the same M13 subclone sequence.

Figure 6

APPROVED	O.G. FIG.	
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Table 2 Primers for PCR amplification of the human *spastin* ORF

Primer set	Forward primer	Reverse primer	Product size (bp)
1 (SEQ ID NO: 21)	CCTCCAGTACTGTGTTATTTGTGAG	CAAGAACTTCCTCAGGGCATC	(SEQ ID NO: 22) 603
2 (SEQ ID NO: 23)	GATGCATCTATACAACATCCGCT	GGTGGAAATAGGTTCTCTTC	(SEQ ID NO: 24) 581
3 (SEQ ID NO: 25)	AAAAATGAGAAATCCAAATGTGCT	GCACTAAGGCTAGGTTTGTGAAG	(SEQ ID NO: 26) 592
4 (SEQ ID NO: 27)	GCTCCTCACTTCCTCTTGTG	CGTGAATTGGCTTCATGATAA	(SEQ ID NO: 28) 602
5 (SEQ ID NO: 29)	AGCAATCAGATTCCAGCAAGC	GATGGGAATGTCAGTGATATGG	(SEQ ID NO: 30) 611
6 (SEQ ID NO: 31)	GGGAGAAAGTTGACAAAGTTGGA	CTTTGGTTTCATCACTGGGAAG	(SEQ ID NO: 32) 624
7 (SEQ ID NO: 33)	TCCAAAGCATTGAACACACCT	CAGGTCCCGTAAGACACTCAG	(SEQ ID NO: 34) 631
8 (SEQ ID NO: 35)	CAATGGGTGCTTTGCTGTTAC	CGAAGAACTCCCGAGAACTCA	(SEQ ID NO: 36) 620
9 (SEQ ID NO: 37)	GCTGGCTGCAAAACAGATACTAC	GCAAAACATGGTTTCAGGCTTA	(SEQ ID NO: 38) 604
10 (SEQ ID NO: 39)	CAACAATCCGCTTCCTTCCAT	ATTATTCGTGCGCAAAAGCTGA	(SEQ ID NO: 40) 651
11 (SEQ ID NO: 41)	TTCCGCGAACTTTTGAAACC	ACACAAAAGTGCTGGCCCTTGC	(SEQ ID NO: 42) 625
12 (SEQ ID NO: 43)	GATGCAAGGCGACAGAAATC	ATACAGCACATTTAGAGCTCCAGT	(SEQ ID NO: 44) 626
13 (SEQ ID NO: 45)	GCATCAGACAGAAATGGTCCAG	GCAATTCAACATATGCAGGAG	(SEQ ID NO: 46) 624
14 (SEQ ID NO: 47)	GTGAATGGCCACTTTTGCACT	TGATATCAGCAGGGGTCAACAT	(SEQ ID NO: 48) 648
15 (SEQ ID NO: 49)	ACCACACGCAAAACAGTAGCA	GCCATGCATTCTTAAGCCAAAG	(SEQ ID NO: 50) 609
16 (SEQ ID NO: 51)	TGACATTTCCAGCTTTTGCTGA	AGCGGCCACTGATGGATTTAT	(SEQ ID NO: 52) 631
17 (SEQ ID NO: 53)	AAATGATTTTGAGGCACCTTTTG	TTCCACCCAGGATGTCATAAA	(SEQ ID NO: 54) 609
18 (SEQ ID NO: 55)	ACAGTAGACTAAAGCAAGCAAAAGC	ATCAAGAGGAGGATCCAGGTT	(SEQ ID NO: 56) 645
19 (SEQ ID NO: 57)	CATCCTGCCCTATTCTTCCAG	TAAAGCGCAAGGTCTCGTACA	(SEQ ID NO: 58) 618
20 (SEQ ID NO: 59)	TGAGGGCAACAATAGATCC	TCTGCTGTGGGAATAGGATT	(SEQ ID NO: 60) 612
21 (SEQ ID NO: 61)	GCAAGCCCTAAGAGAAGGATT	TGCTTTGAGAGCTTTCTCTCAG	(SEQ ID NO: 62) 647
22 (SEQ ID NO: 63)	TGAAAGAGAAAGATGCTGACAATTC	GTAAGTCTGTCCGGCTGAAGG	(SEQ ID NO: 64) 654
23 (SEQ ID NO: 66)	CATCCCATTTCAGTCAGACA	TTCGTGCTACAAACACATTCAAGA	(SEQ ID NO: 66) 638

Figure 7



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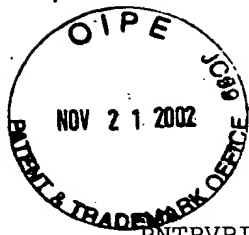
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O.G. FIG.		SUBCLASS	
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LOCUS AF193557 11493 bp DNA ROD
DEFINITION Mus musculus saccin gene, complete cds.
ACCESSION AF193557
VERSION AF193557.1 GI:6907043
KEYWORDS .
SOURCE house mouse.
ORGANISM Mus musculus
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
Mammalia; Eutheria; Rodentia; Sciurognathi; Muridae; Murinae; Mus.
REFERENCE 1 (bases 1 to 11493)
AUTHORS Engert, J.C., Berube, P., Mercier, J., Dore, C., Lepage, P., Ge, B.,
Bouchard, J.P., Mathieu, J., Melancon, S.B., Schalling, M.,
Lander, E.S., Morgan, K., Hudson, T.J. and Richter, A.
TITLE ARSACS, a spastic ataxia common in northeastern Quebec, is caused
by mutations in a new gene encoding an 11.5-kb ORF
JOURNAL Nat. Genet. 24 (2), 120-125 (2000)
MEDLINE 20120709
REFERENCE 2 (bases 1 to 11493)
AUTHORS Engert, J.C., Berube, P., Dore, C., Lepage, P., Ge, B., Hudson, T.J. and
Richter, A.
TITLE Direct Submission
JOURNAL Submitted (08-OCT-1999) Genome Centre, Montreal General Hospital,
1650 Cedar Ave., Montreal, QC H3G 1A4, Canada
FEATURES Location/Qualifiers
source 1..11493
/organism="Mus musculus"
/db_xref="taxon:10090"
mRNA <1..>11493
/product="saccin"
CDS 1..11493
/note="molecular chaperone"
/codon_start=1
/product="saccin"
/protein_id="AAAF31263.1"
/db_xref="GI:6907044"

/translation="MNTFWPGRELVVQWYPFSEDKRHPSLSWLKMVWKNLYIHFSEDL
TLFDEMPLIPRTLLNEDQTCVELIRLRIPSVVILDDTEAQLPEFLADIVQKLGIVL
KRLDTSIQHPLVKKYIHSPLPSAILQIMEKIPLQKLCNQIASLLPTHKDALKFLASL
TDTSEKEKRIIQELTIFKRINHSSDQGISSYTKLKGCKVLDHTAKLPTDLRLSVSVID
SSDEATIRLANMLKIEKLKTTSCLEKLVLDIGNAFYTQEEVTQLMLWILENLSSLKNE
NSNVLDWLMPLKFIHMSQGHVVAAGDLDFDPDIEVLRDLFYNEEEACFPPTIFTSPDIL
HSLRQIGLKNESLKEKDVVQVARKIEALQVSSCQNDVLMKKAKTLLLVLNKNQTLL
QSSEGKMAKKIKWVPACKERPPNYPGLSVWKGDLNLCAPPDMCDAAHAVLVGSSLP
LVESVHVNLEQALSIFTKPTINAVLKHFKTVVDWYTSKTFSEDEYYQFQHILLEIYGF
MHDHLSEGKDSFKALKFPWVWTGKNFCPLAQAVIKPTHDLDLQPYLYNVPKTMKAFHQ
LFKACGSIEELTSDHISMVIQKVKYLSKQELSEESKQNLHMLNIMRWLYSNQIPAS

Figure 8A



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PNTVPPIYHSRNP SKLVMKPIHECCYCDIKVDDLNDLLED SVEPIILVHEDI PMKTAE
WLKVPCLSTRLINPENMGFEQSGQREPLTVRIKNILEEYPSVSDIFKELLQONADDANA
TECSFMIDMRRNMDIRENLLDPGMAACHGPALWSFNNSEFSDSDFLNITRLGESLKR
EVDKVGKFGFGFNSVYHITDIP IIMSREFMIMFDPNINHISKHIKDRSNPGIKINWSK
QQKRLRKFPNQFKPFIDVFGCQLPLAVEAPYSYNGTLFRLSFRTQQEAKVSEVSSTCY
NTADIYSLVDEFSLCGHRLLIFTQSVNSMYLKYLKIEETNP SLAQDTIIKKKVCPSK
ALNAPVLSVLKEAAKLMKTCSSSNKKLPTDVPKSSCILQITVEEFHHVFRRRIADLQSP
LFRGPDDDPATLFEMAKSGQSKKPSDEL POKTVDCTTWLICTCMDTGEALKFSLNESG
RRLGLVPCGAVGVLLHETQE QKWTVKPHIGEVFCYLPLRIKTGLPIHINGCFAVTSNR
KEIWKTDTKGRWNTTFMRHVIVKAYLQALS VLRLDAIGGELTDYTYAVWPD PDLVHD
DFS VICKGFYEDIAHGKGKELTRVFS DSGSMVWSMKNVRFLDD SILQRKDVGSAAFKIF
LKYLKKTGSKNLCAVELPSSVKAGFEEAGCKQILLENTFSEKQFFSEVFFPNIQIEIA
ELRDPLMNFVLNEKLDEFSGILRVTPCVPCSLEGHPLVLP SRLIHPEGRVAKLFDTKD
GRFPYGSTQDYLNPIILIKLVQLGMAKDDILWDDMLERAESVAEINKSDHAAACLRS
ILLSLIDEKLKIKDPRAKDFAAKYQTI PFLPFLTKPAGFSLEWKGN SFKPETMFAATD
IYTA EYQDIVCLLQPI LNENSHSFRGCGSVSLAVKEFLGLLKKPTVDLVINQLKQVAK
SVDDGITLYQENITNACYKYLHEAVLQ NEMAKATIEKLKPF CFI LVENVYVESEKVS
FHLNFEAAPYLYQLPNKYKNFREL FESVGVRSFTVEDFALVLESIDQERGKKQITE
ENFQLCRRIISEGIW SLIREKRQEFCEKNY GKIILLPD TNLLLLPAKSLCYND CPWIKV
KDSTVKYCHADIPREVA VKLGAIPKRHKALERYASNICFTALGTEFGQKEKLTSRIKS
ILNAYPSEKEMLKELLQONADDAKATEICFVFDPRQHPVDRI FDDKWAPLQGPALCVYN
NQPFTEDDVRGIQNLGKGTKEGNPCKTGHYGIGFNSVYHITDCPSFISGNDILGIFDP
HARYAPGATSVSPGRMFRDL DADFRTQFSDVLDLYLGNHFKLDNCTMFRFPLRNAEMA
QVSEISSVPSSDRMVQNLLDKLRSDGAELLMFLNHMEKISICEIDKATGGLNVLYSVK
GKITDGDRLKRKQFHASVIDSVTKRQLKDIPVQQITYTMDTEDSEGNLTTWLICNRS
GFSSMEKVS KSVISAHKNQDITLFRGGV AACITHNYKKPHRAFCFLPLSLETGLPFH
VNGHFALDSARRNLWRDDNGVGVRS DWNNSLMTAL IAPAYVELLIQLKKRYFPGSDPT
LSVLQNTPIHVVKDTLKKFLSFFPVNRLDLQPDLYCLVKALYSCIHEDMKRLLPVVRA

APPROVED	O.G. FIG.	
	CLASS	SUBCLASS
BY	DRAFTSMAN	

Figure 8B



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

PNIDGSDLHSAVITWINMSTSNKTRPFFDNLLQDELQHLKNADYNITTRKTVAENVY
RLKHLLEIGFNLVYNCDATANLYHCLVDADIPVSYVTPADVRSFLMTFSSPDTNCHI
GKLPCRLOQTNLKLFHSLKLLVDYCFKDAEESFEVEGLPLLITLDSVLQIFDGKRPK
FLTTHYHELIPSRKDLFMTNTLYLKYSSVLLNCKVAKVFDISSFADLLSSVLPREYKTKN
CAKWKDNFASWLNKNAWHFISESVSVTDDQEEPKPAFDVIVDILKDWALLPGTKFTV
STSQLVVPEGDVLIPLSLMHIAVFPNAQSDKVFHALMKAGCIQLALNKICSKDSALVP
LLSCHTANIDSPASILKAVHYMVQTSTFRTEKLMENDFEALLMYFNCNLSHLMSQDDI
KILKSLPCYKSIISGRYMSIAKFGTCYVLTKSIPSAEVEKWTQSSSSAFLEEKVHLKEL
YEVLCGPVDDLEVYLKHLPLKIENLSYDAKLEHLIYLNRLASIEEPSEIKEQLFEK
LESLLIHDANNRLKQAKHFYDRTVRVFEVMLPEKLFIPKEFFKKLEQVIKPNQAAF
MTSWVEFLRNIGLKYALSQQQLLQFAKEISVRANTENWSKETLQSTVDILLHHIFQER
MDLLSGNFKELSLIPFLCERAPAEYIRFHPQYQEVNGTLPLIKFNGAQVNPKEKQC
DVLQLLWTSCPIPEKATPLSIKEQEGSDLAPQEQLQVLNMLNVNLDPPDKVINNC
RNICNITTLDEEMVKTRAKVLRSIYEFLSAEKREFRFQLRGVAFVMVEDGWKLLKPEE
VVINLEYEADFKPYLYKLPLELGTFFHQLFKHLGTEDIISTKQYVEVLSRIFKSSEKQ
LDPNEMRTVKRVVSGLFKSLQNDVSVKVRSDLENARDLALYLPQDGLVKSSILVDDD
APHYKSRIQGNIGVQMLVDLSQCYLGKDHGFHTKLIMLFPQKLRPRLLSSILEEQDE
ETPKVCQFGALCSLQGRQLLLSSEQFITGLIRIMKHENDNAFLANEKKAIRLCKALR
EGLKVSCEFELQTTLRVKGFNPIPHSRSETFAFLKRFGNAVILLYIQHSDSKDINFL
ALAMTLKSATDNLISDTSYLIAMLGENDIYRISEKLDLGVKYDSSEPSKLELPMPGT
PIPAEIHYTELLMDPMNVFYPGEYVGYLVDAEGGDIYGSYQPTYTYAIIVQEVEREDAD
NTSFLGKIYQIDIGYSEYKIVSSLDLYKFSRPDESSQNRDSAPTTPTSPTEFLTPGLR
SIPPLFSGKESHKSPSTKHHSPRKLKVNALPEILKEVTSVVEQAWKLPESEKKIIRR
LYLKWHDPKNPENHDIANEVFKHLQNEINRLEKQAFLDQADRASRRTFSTSASRFQS
DKYSFQRFYTSWNQEATSHKSERQQQSKEKCPPSAGQTYSQRFVPPPTFKSVGNPVEA
RRWLRQARANFSAARNDLHKNANEWVCFKCYLSTKLALIAADYAVRGKSDKDVKPTAL
AQKIEEYSQGLEGLTNDVHTLEAYGVDSLKTRYPDLLPFPQIPNDRFTSEVAMRVMEC
TACIIIKLENFIQKV"

Figure 8C



App No.: 09/693,205
Title: Identification of ARSACS
Inventors: Thomas J. Hudson, et al.

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BASE COUNT 3599 a 2281 c 2387 g 3226 t
ORIGIN

APPROVED	O.G. FIG.		SUBCLASS
	CLASS		
BY			
DRAFTSMAN			

1	atgaatacat	tctggccttg	tgcagagttg	gtgggttcagt	ggatccatt	tagtgaagac
61	aaacgtcacc	catccctttc	atggccttaag	atgggtttgga	agaatctcta	tatacatttc
121	tcggaagatt	tgacttttatt	tgatgagatg	ccacttatcc	ctagaactct	actgaatgag
181	gaccagacgt	gtgtggaact	catcagactc	aggatcccat	cagtagtcat	tttagatgat
241	gaaactgaag	ctcagctttcc	agaattctta	gcagatattg	tacaaaaact	tggagggatt
301	gtcctgaaaa	gactagatac	ctctattcag	catccacttg	ttaaaaata	cattcattcc
361	ccactccga	gtgctatttt	gcagataatg	gagaagatac	ctctacagaa	gttgtgtaat
421	caaatacat	cattactttcc	aaccacaaaa	gatgctctaa	ggaagttttt	ggccagctta
481	actgatacca	gtgaaaaaga	gaaaaagaata	attcaagaat	tgacaatatt	caaaaagaatt
541	aatcactcat	cagatcaagg	gatttctctc	tacacaaaa	taaaaggatg	taaagttttg
601	gatcataccg	ccaagctttcc	aacagatcta	cggctatcag	tttcagtaat	agatagtagt
661	gatgaagcca	ccattcggtt	ggcaaacatg	ttgaaaattg	aaaaattgaa	gactacaagc
721	tgtttaaaagt	ttgttttaaa	agatattgga	aatgcatttt	atacacagga	agaggttaaca
781	caacttatgc	tttggtacct	tgagaatcta	tctctcttta	aaaatgagaa	ttcaaatgtg
841	cttgattggt	taatgccact	aaaattcatt	catatgtccc	agggacatgt	ggtagcagct
901	ggtgatctct	ttgatcctga	tatagaagta	ctaagggatc	tcttttataa	tgaagaagaa
961	gcttgtttcc	cacctacaat	ttttacctca	ccagatatcc	ttcactcttt	gagacagatt
1021	ggcttaaaaa	atgaatccag	tctaaaagaa	aaagatgttg	tacaagtggc	aagaaaaatt
1081	gaagctttac	aggtcagttc	ctgtcagaat	caggatgttc	tcatgaagaa	agccaaaaca
1141	ctcttactgg	tcttgaataa	aaaccagaca	ctcttgacgt	cttctgaagg	gaagatggca
1201	ttgaagaaaa	tcaaattgggt	tccagcctgc	aaggaaaagac	ctccaaatta	tcccgggtcc
1261	ttagtctgga	aaggggatct	ctgtaatctt	tgtgcacctc	cagatagtgt	tgatgcggca
1321	catgcagttc	tagtaggctc	ctcacttctc	cttggttgaa	gtgtccatgt	gaacctggag
1381	caggcgtcca	gcatcttcac	aaagcctact	atcaatgctg	tcttaaaaca	ctttaaact
1441	gttgttgact	ggtatacttc	aaaaaccttt	agtgatgaag	attactatca	gttccaacat
1501	atthttgctg	aaatttatgg	gttcatgcat	gatcatctga	gtgaagggaa	ggattctttt
1561	aaagccttga	agtttccatg	ggtttggtac	ggcaaaaaact	tttgctctct	tgcccaggct
1621	gtgataaagc	caaccatgat	tctggatctt	cagccttatt	tataaatgt	gcctaaaacc
1681	atggcaaaat	tccaccagtc	gttcaaaggc	tgtgggtcaa	tagaagagtt	gacatcagat
1741	catatttcca	tggtcattca	gaaagtttat	ctcaaaaagt	accaggagtt	gagtgaagaa
1801	gaaagtaaac	aaaatcttca	tctcatgttg	aatattatga	gatggctcta	tagcaatcag
1861	attccagcaa	gccctaatac	accagttcct	atthtatcaca	gcagaaatcc	ttccaaactt
1921	gtcatgaagc	caattcatga	atgctgttat	tgtgacatca	aagttgatga	cctcaatgac
1981	ttgcttgaag	attcagtggg	accaattatc	ttggtacatg	aagatatacc	catgaaaact
2041	gacaagggtg	tgaaagtctc	gtgccttagt	acaagactga	tcaatctga	aaacatgggg
2101	tttgagcagt	cagggcaaa	agagcctctt	actgtaagga	ttaaaaatat	tttggaagaa
2161	tacccttccg	tgacagatat	ttttaaagag	ctacttcaaa	atgctgatga	tgcaaatgcc
2221	acagaatgca	gcttcatgat	tgatatgaga	aggaatatgg	acatacggga	aatctctctg
2281	gaccagggga	tggcagcttg	tcatggacct	gctctgtggt	cattcaacaa	ttctgaattc
2341	tcagatttcag	atttcttaaa	cataacgagg	ttaggagagt	ctttaaaaag	gggagaagtt
2401	gacaagggtg	ggaaattttg	tcttggtttt	aattctgtgt	accacatcac	tgacattccc
2461	atcattatga	gcagagaatt	tatgataatg	tttgatccaa	acataaatca	tatcagcaaa
2521	cacattaaag	atagatcgaa	tcctggaatc	aaaattaatt	ggagtaagca	gcagaaaaga
2581	cttaggaagt	tccccaacca	gttcaaacca	tttatagatg	tatttggtctg	tcagttacct
2641	ttggctgttg	aagctcctta	cagctacaat	ggaactcttt	tccgactgtc	ctttagaaca
2701	cagcaggaag	caaaagttag	tgaagtttag	agtacttgct	acaatactgc	ggatattttac
2761	tccctagtgg	atgaatttag	tctttgtggg	cacagactta	tcattttttac	tcagagtgtg
2821	aactcgatgt	atttgaaata	cttgaaaatt	gaagaaacca	atcctagctt	agcacaagat
2881	acaatcataa	ttaagaaaaa	agtttgcccc	tccaaagcat	tgaatgcacc	agttttaagt
2941	gttttaaaag	aagctgctaa	actcatgaag	acttgtagca	gcagcaacaa	gaagcttccc
3001	acggatgtgc	caaagtcatc	ttgcattctt	cagatcacag	tcgaagaatt	ccaccatgtg
3061	tttaggagga	ttgctgactt	acagtcacca	ctatttcgag	gtccagatga	gtcccagctc
3121	actctctttg	aaatggctaa	atctggccaa	tcaaaaaagc	catcagatga	gttgccacaa
3181	aagacagtag	attgtaccac	atggcttata	tgcacatgca	tggatacagg	agaagctctc
3241	aagttttcct	tgaatgaaag	tgaagaagaa	ttagggctgg	ttccttgtgg	ggcagtaggg

Figure 8D



App No.:
Title:
Inventors:

09/693,205
Identification of ARSACS ...
Thomas J. Hudson, et al.

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APPROVED	O.G. FIG.	
	CLASS	SUBCLASS
BY	DRAFTSMAN	

3301 gttctcttgc atgaaaccca ggaacagaag tggaccgtga aaccacacat aggagaagtg
3361 ttttgcctatt tacctctacg aatcaaaaca ggggttgccaa ttcacatcaa tgggtgcttt
3421 gctgttactt caaataggaa agaaatctgg aagacagata caaaaggctg atggaatacc
3481 acattcatga ggcattgcat tgtgaaagct tacttacaag ccctcagtg cttacgggac
3541 ctagccattg gtggtgagct gactgattat acttactatg cagtgtggcc tgatcctgat
3601 ctagtctatg atgacttctc tgtgatctgt aaaggatttt atgaagacat tgctcatggg
3661 aaggggaagg agttgaccag agtcttctct gatgggtcta tgtgggtttc catgaagaat
3721 gtgaggtttc tggatgactc tatacttcaa aggaaagatg ttgggttcagc agccttcaag
3781 atatttctga agtacctcaa gaaaacagga tccaaaaacc tctgtgctgt tgagcttcct
3841 tcttcagtaa aagcaggatt tgaagaggct ggctgtaaac agatactgct ggaaaataca
3901 ttttcagaga aacagttctt ttcagaagtc ttctttccta atatccagga aattgaagca
3961 gaacttagag atcctctgat gaattttgtc ctaaatgaaa aacttgatga gttctcagga
4021 attcttcgtg ttacctcttg tgttcttgc tcttgaggg gccatccttt ggttttgcct
4081 tcaagattga tccatcctga aggacgagtt gcaaagttat ttgatactaa agatggaaga
4141 ttcccttatg gttccacaca ggattacctc aatcctatta tcttgattaa gctcgttcag
4201 ttaggcattg caaaagatga tattttgtgg gatgacatgc tagagcgtgc agagtctgta
4261 gctgagatta ataaaagtga ccatgctgct gctgtcttaa ggagtagtat tctgctaagc
4321 cttattgatg agaagctaaa aataaaggat cctagagcaa aggattttgc tgcaaatat
4381 caaacaattc ctttctctcc atttctaaca aagccagcag gtttttcttt agaatggaaa
4441 gggaacagct ttaagcctga aacctggtt gcagcaactg acatttacac agctgaatat
4501 caagatatag tctgtctttt gcaaccaatt cttaatgaaa attcccattc ctttagaggc
4561 tgtggttcag tgtctttggc tgtaaggag tttttgggtt tactaaagaa gccaacagtt
4621 gatctggtaa taaaccagtt gaagcaagtt gcaaaatcag ttgatgatgg cattacattg
4681 taccaggaaa atatcaccaa cgcttgctac aaataacctc atgaagcag atgacagaat
4741 gaaatggcca aggcaacaat tattgagaag ctaaagccat tttgtttcat tctagttgag
4801 aatgtatatg ttgagtcaga aaaggtttct ttctacttga actttgaagc agcaccatac
4861 ctttatcagt tacctaacaa gtataaaaaat aatttccgtg agctttttga aagtgtgggt
4921 gtgcgacagt catttactgt tgaagacttt gccctagttt tggagtctat tgatcaagag
4981 agaggaaaaa aacaaataac agaagagaat ttctagcttt gccgacgaat aatcagtgaa
5041 ggcatctgga gtctcattga agaaaagaga caagaatttt gtgagaaaaa ttatggcaaa
5101 atattactgc cagacactaa cctgctgctg ctccctgcta agtcattatg ctacaatgac
5161 tgtccctgga taaaagtaaa ggactccact gtcaagtatt gccatgccga cataccccg
5221 gaagtagctg taaaacttgg tgcaatacca aagagacata aagcattaga aagatatgca
5281 tccaacatct gtttcacagc tctaggtaca gaatttgggc agaaaagaaa actgaccagc
5341 agaattaaga gcatttctca tgcctatcct tcagaaaagg aaatgctgaa agagcttctt
5401 caaatgctg atgatgcaaa ggccacagag atctgctttg tgttctgacc tagacagcat
5461 cctgttgacc gaatatttga tgataagtgg gccccactgc aagggccagc actgtgtgtt
5521 tacaacaacc agccatttac agaagatgat gttagaggaa ttcagaatct tgggaaaggc
5581 accaaagaag ggaatccttg caaaacagga cattatggaa tccgattcaa ttccgtttat
5641 catattacag actgcccttc ttttatttct ggcaatgaca tcttggttat ttttgatccc
5701 catgccagat atgcaccagg agccacatca gttagcctg gacgcattgt tagagattg
5761 gatgcagact ttagaaccca gttctcagat gttctagatc tgacttggg aaaccacttt
5821 aaactggaca attgtacaat gtttagattt cctctgcgta atgcagagat ggcacaagtt
5881 tcagaaattt cttccgttcc atcatcagac agaatggctc agaatctttt ggacaagtta
5941 cggctctgat gggcagaact tctaattgtt ctcaaccaca tggagaaaat atctatttgt
6001 gaaatagata aggccacagg aggtctgaat gtgctctatt cagtaaaagg caagatcact
6061 gatggagacc gattgaaaag gaagcaattc cacgcctctg taattgacag tgttactaaa
6121 aagagacagc tcaaggacat accagttcaa caaataacct acactatgga tactgaggat
6181 tctgaaggaa atctgaccac atggctcatc tgtaatagat caggattttc aagtatggaa
6241 aaagtatcca agagtgtaat atcagctcac aagaaccaag atatcaccct tttcccacgt
6301 ggtggagtag cagcctgcat tactcacaat tataaaaagc cccacagagc cttctgcttt
6361 ctgcctctct ctttgagagc agggctgcca ttctcatgtga atggccactt tgctctagat
6421 tcagccagaa gaaacttgtg gcgtgatgat aatggggttg gtgttcgaag tgactggaat
6481 aatagtttaa tgacagcatt aatagcacct gcatatgttg agttactaat ccagttaaaa
6541 aaacggtatt tccctggttc tgacccaaca ttatcagttt tacagaacac acccattcat
6601 gtcgtaaagg acacattaaa gaagtttctg tcttctttc cagttaacag gctggatctg
6661 cagccggact tatattgctt agtaaaagca ctttacagtt gcattcatga agacatgaag

Figure 8E



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Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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APPROVED BY DRAFTSMAN	O. G. FIG.	CLASS	SUBCLASS

6721	cgtctttttgc	ctgtttgttcg	ggctccaaat	attgatggct	cagatttgca	ctctgcagtc
6781	ataattactt	ggatcaatat	gtctacttca	aataaaaacta	gaccattttt	tgataactta
6841	ctacaggatg	aattacagca	ccttaaaaaat	gcagattata	acatcacaaac	tcgaaaaaca
6901	gtcgcagaga	atgtctacag	actgaagcac	ctgctcttag	aaattgggtt	caacttgggt
6961	tataactgtg	atgaaaactgc	taacctttac	cattgccttg	tagatgcaga	tatccctgtc
7021	agctatgtga	ctcctgctga	tgttaggtcc	ttcttaataga	ctttctcttc	tcctgacact
7081	aattgccata	ttgggaagct	gccttgctcg	cttcagcaga	ctaacctaaa	actttttcac
7141	agtttaaaac	tttttagttga	ttactgtttt	aaagatgctg	aagaaagtga	gtttgaagtt
7201	gagggactgc	ccctactcat	tacactggac	agtgtcttgc	agatttttga	tggtaaacga
7261	cccaagtttc	taacaacata	ccatgaatta	attccatcgc	gtaaagactt	gtttatgaac
7321	accttatact	tgaaatacag	tagtgttttg	ttgaactgca	aagttgcaa	agtgtttgac
7381	atttccagct	ttgctgactt	actctcttct	gtgttgccctc	gtgagtacaa	gaccaaaaaac
7441	tgtgcaaagt	ggaaagacaa	ttttgccagt	gaatcttggc	ttaagaacgc	atggcatttt
7501	atcagtgaat	cagtaagtgt	aacggatgat	caggaagaac	caaagccagc	atttgatgtc
7561	attgttgaca	tccttaaaga	ctgggcattg	cttccaggaa	caaagttcac	tgtgtcaacc
7621	agtcagcttg	tgggttcctga	gggagacgtg	ttgattcccc	tgagcctcat	gcacattgct
7681	gtgttcccaa	atgctcagag	tgataaggtt	tttcacgctc	tgatgaaagc	tggctgtatt
7741	cagctggctt	tgaacaaaaat	ctgctctaaa	gacagcgcac	tagttcctct	gtgtgcatgc
7801	cacacagcaa	acatagatag	ccctgcaagc	atcttgaagg	ctgtgcatta	tatggttcag
7861	acgtcaacat	ttagaactga	aaaactaatg	gaaaatgact	ttgaagcact	tttgatgtat
7921	ttcaactgta	atttgagtca	cttgatgtcc	caagatgaca	taaaaatttt	aaagtccctc
7981	ccatgctaca	aatccatcag	tggccgctat	atgagcattg	caaaatttgg	aacgtgctat
8041	gtgcttacca	aaagtattcc	ttcagctgaa	gtggaaaaat	ggacacagtc	atcctcttcc
8101	gcgtttcttg	aagaaaaaggt	gcatttataaa	gaactctatg	aggtgcttgg	ctgtgtgcca
8161	gtagatgac	tggaggtgta	tttgaacat	cttctgcca	aaattgaaaa	tctctcttat
8221	gatgcaaagt	tggagcacct	gatttatctg	aagaatagac	tggcaagcat	cgaggaaccg
8281	tcagagatta	aggagcaact	ttttgaaaaa	ctggaaagct	tattgattat	ccacgatgcc
8341	aacaatcgac	taaagcaagc	aaaacatttc	tatgacagaa	ctgtgagagt	ttttgaagtt
8401	atgcttcttg	aaaaattggt	tattcctaag	gagttcttta	aaaaatttga	acaagtaatc
8461	aaacctaaaa	atcaagctgc	atttatgacg	tctctgggtg	aattatttga	aaattatttga
8521	ctgaagtaag	cgctctccca	gcagcagttg	ttacagtttg	ccaaggaaat	cagtgtgagg
8581	gcaaatcacg	aaaactggtc	taaagaaacc	ctgcaaagta	cagttgacat	ccttctccat
8641	cacatattcc	aagaacgaat	ggatttggtta	tctggaaatt	ttctgaaaga	actgtcctta
8701	ataccattct	tgtgtcctga	acgggcccc	gctgagtaca	ttcggtttca	ccctcagtac
8761	caggaggtaa	acggaacact	tcctcttata	aagttcaatg	gagcacaaat	gaatccaaag
8821	ttcaagcaat	gtgatgtact	ccagctgtcg	ttgacatctt	gcctattctt	tcagagaaa
8881	gccacaccgt	tgagcattaa	agaacaagaa	ggcagtgacc	tcgctccaca	ggaacagctt
8941	gaacaagttt	taaatatgct	taatgttaac	ctggaccccc	ctcttgataa	ggtcattaat
9001	aattgcagaa	acatatgcaa	cataacaact	ttggatgagg	aaatggtaaa	aactagagca
9061	aaggctcctaa	ggagcatata	tgaatttctg	agtgcagaaa	aacgagagtt	ccgttttcag
9121	cttcgggggtg	tggcctttgt	aatggtagaa	gacggatgga	aacttctgaa	gcctgaggaa
9181	gtagtataa	acctggagta	tgaggctgat	tttaaacctt	atctgtacaa	gctgccttta
9241	gagcttgga	cttttcatca	gctgttcaaa	catttaggta	ctgaagatat	catttccact
9301	aagcaatatg	ttgaagtgtt	aagccgaata	ttcaaaagct	ctgaaggaaa	gcagctagac
9361	cctaatagaaa	tgcgtacagt	taagagagtg	gtttctggcc	tattcaagag	tctacaaaat
9421	gattcagtea	aggtgaggag	tgacctggag	aatgcccggg	acctcgcact	ctaccttcca
9481	agccaggatg	ggaagtgggt	gaagtcaagc	atcttgggtg	tcgatgatgc	gccacattat
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9601	ttagggaag	accatggatt	tcacactaag	ctgataatgc	tctttcctca	aaagcttcga
9661	cctcgtctgc	tgagcagtat	acttgaagag	cagcttgatg	aggagacccc	taaagtgtgc
9721	cagtttggcg	cattgtgtctc	tcttcaggga	agactgcagc	ttctcttgct	ttcagagcag
9781	ttcatcacag	gactcattcg	aatcatgaag	catgaaaatg	ataatgcttt	cctggccaat
9841	gaagaaaaag	ccataagact	ttgcaaagct	ctaagagaag	ggctgaaagt	ttcctgtttt
9901	gagaagcttc	agacaacatt	aagggtttaa	ggttttaatc	ctattcccca	tagcaggagt
9961	gaaactttcg	cttttctaaa	gcgatttggc	aatgcagtca	tcttgctcta	catccaacat
10021	tcagacagca	aagacattaa	ctttctgcta	gccttagcga	tgacacttaa	atcagcaact
10081	gacaatttga	tttctgacac	gtcatactta	attgctatgc	tgggatgcaa	tgacatttac

Figure 8F



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Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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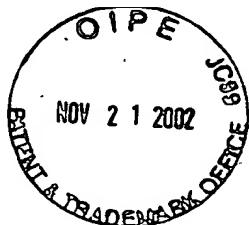
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APPROVED BY DRAFTSMAN	O.G. FIG.	CLASS	SUBCLASS

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10621 accaaacacc attccccag aaagctcaag gtgaatgctt taccagaaat cttaaaagaa
10681 gtgacatcag tgggtggagca agcttggaag cttccagaat cagagcggaa aaagatcatt
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11461 aaacttgaaa attttatata acagaaggtg tga

//

Figure 8G



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, *et al.*

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LOCUS AF193556 12793 bp DNA PRI 07-FEB-2000

DEFINITION Homo sapiens saccin (SACS) gene, complete cds.

ACCESSION AF193556 VERSION AF193556.1 GI:6907041KEYWORDS .

SOURCE human.

ORGANISM Homo sapiens Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.

REFERENCE 1 (bases 1 to 12793) AUTHORS Engert,J.C., Berube,P., Mercier,J., Dore,C., Lepage,P., Ge,B., Bouchard,J.P., Mathieu,J., Melancon,S.B., Schalling,M., Lander,E.S., Morgan,K., Hudson,T.J. and Richter,A. TITLE ARSACS, a spastic ataxia common in northeastern Quebec, is caused by mutations in a new gene encoding an 11.5-kb ORF JOURNAL Nat. Genet. 24 (2), 120-125 (2000) MEDLINE 20120709

REFERENCE 2 (bases 1 to 12793) AUTHORS Engert,J.C., Berube,P., Dore,C., Lepage,P., Ge,B., Hudson,T.J. and Richter,A. TITLE Direct Submission JOURNAL Submitted (08-OCT-1999) Genome Centre, Montreal General Hospital, 1650 Cedar Ave., Montreal, QC H3G 1A4, CanadaFEATURES Location/Qualifiers source 1..12793 /organism="Homo sapiens" /db_xref="taxon:9606" /chromosome="13" /map="between D13S232 and D13S292" mRNA 1..12793 /gene="SACS" /product="saccin" gene 1..12793 /gene="SACS" CDS 77..11566 /gene="SACS" /note="molecular chaperone" /codon_start=1 /product="saccin" /protein_id="AAF31262.1" /db_xref="GI:6907042" /translation="

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QSSEGKMTLKKIKWVPACKERPPNYPGSLVWKGDLNLCAPPDMCDVGHAILIGSSLP
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LFKVCGSIEELTSDHISMVIQKIYKSDQDLSEQESKQNLHMLNIIRWLYSNQIPAS
PNTVPPIHHSKNPSKLIMKPIHECCYCDIKVDDLNDLLEDSEVEPIILVHEDIPMKTAE
WLKVPCLSTRLINPENMGFEQSGQREPLTVRIKNILEEYPSVSDIFKELLQNADDANA
TECSFLIDMRRNMDIRENLLDPGMAACHGPALWSFNNSQFSDSDFVNITRLGESLKR
EVDKVGKFGLGFNVSYYHITDIPIIMSREFMIMFDPNINHISKHIKDKSNPGIKINWSK
QQKRLRKFPNQFKPFIDVFGCQLPLTVEAPYSYNGTLFRLSFRTQGEAKVSEVSSTCY
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LFRGPDDDDPAALFEMAKSGQSKKPSDELSQKTVECTTWLLCTCMDTGEALKFSLSESG
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Figure 9A



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

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GKITDGDRLKRKQFHASVIDSVTKKRQLKDIPVQQITYTMDTEDSEGNLT TWLICNRS
GFSSMEKVS KSVISAHKNQDITLFRGGVAACITHNYKKPHRAF CFLPLSLETGLPFH
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BASE COUNT 4163 a 2256 c 2487 g 3887 tORIGIN
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Figure 9B



App No.: 09/693,205
Title: Identification of ARSACS . . .
Inventors: Thomas J. Hudson, et al.

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O.G. FIG.	SUBCLASS	
	CLASS	
APPROVED BY	DRAFTSMAN	

tgagaatcca aatgtgcttg agtgggttaac accattaaaa ttcattccaga tatcacagga
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Figure 9C



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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

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cttttcaagt	atggagaaa	tatctaaaa	tgatcatatca	gtcacaaga	accaagatat
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Figure 9D



App No.: 09/693,205
Title: Identification of ARSACS ...
Inventors: Thomas J. Hudson, et al.

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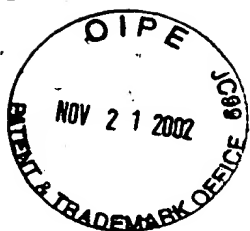
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TECH CENTER 1600/2900

APPROVED BY DRAFTSMAN	O.G. FIG.
	CLASS SUBCLASS

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gaatgcatgg	cattttatta	gtgaatctgt	aagtgtgaaa	gaagatcagg	aagaaacaaa
accaacattt	gacattgttg	ttgatactct	aaaagactgg	gcattgcttc	caggaacaaa
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gaaagccggc	tgtattcagc	ttgctttgaa	caaaatctgt	tccaaagaca	gtgcatttgt
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acaatcatca	tcactctgat	ttcttgaaga	aaaaatacac	ttaaaagaac	tatatgaggt
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tctcagccag	tgctacttag	ggaaagacca	tggatttcac	actaagttga	taatgctctt
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gactcccaaa	gtttgtcagt	ttggagcggt	gtgttctctt	caaggaagat	tgcaagtact
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tgcttttctg	gccaatgaag	aaaaagccat	aagactttgc	aaagccctaa	gagaaggatt
gaaagtatcc	tgctttgaaa	agcttcaaac	aacattaaga	gttaaagggt	ttaatcctat
tccccacagc	agaagtgaag	cttttgcttt	tttgaagcga	tttggtaatg	cagtcactct
gctctacatt	caacattcag	acagtaaaga	cattaatttc	ctgtagcac	tggcaatgac
tcttaaatca	gcaactgaca	atttgatttc	tgacacttca	tatttaattg	ctatgctagg
atgcaatgat	atttacagga	ttggtgagaa	acttgacagt	ttaggagtga	aatatgactc
ttcggagcca	tcaaaactgg	aacttccaat	gcctggcaca	ccaattcctg	ctgaaattca
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tattgtacaa	gaagttgaaa	gagaagatgc	tgacaattct	agttttctag	gaaagatata
tcagatagat	attggttata	gtgaatataa	aatagttagc	tctcttgatc	tgtataagtt
ttcaagacct	gaggaaagct	ctcaaagcag	ggacagtgtc	ccttctacac	caaccagccc

Figure 9E



App No.:
Title:
Inventors:

09/693,205
Identification of ARSACS ...
Thomas J. Hudson, *et al.*

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APPROVED BY DRAFTSMAN	O.G. FIG.	SUBCLASS
	CLASS	

cactgagttc ctcacccctg gcctgagaag cattcctcct cttttctctg gtagagagag
ccacaagact tcttccaaac atcagtcctc caaaaagctt aagggttaatt ctttaccaga
aatcttaaaa gaagtgcacat ctgtggtgga gcaagcatgg aagcttccag aatcggaacg
aaaaaagatt attaggcggt tgtatttgaa atggcatcct gacaaaaatc cagagaacca
tgacattgcc aatgaagttt ttaaaccatt gcagaatgaa atcaacagat tagaaaaaca
ggcttttcta gatcaaaatg cagacagggc ctccagacga acattttcaa cctcagcatc
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acagacttac tctcaaaggt tctttgttcc tcccactttc aagtcggttg gcaatccagt
ggaagcacgc agatggctaa gacaagccag agcaaaactc tcagctgcca ggaatgacct
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cattttattg ctt

Figure 9F